



dbSNP: Database for Short Genetic Variations

Catalog of nucleotide changes for human and other model organisms

<https://www.ncbi.nlm.nih.gov/snp/>

National Center for Biotechnology Information • National Library of Medicine • National Institutes of Health • Department of Health and Human Services

Scope and Access

The NCBI Short Genetic Variations database (dbSNP [1]), commonly known as dbSNP, catalogs short variations in nucleotide sequences for human. These variations include single nucleotide variations, short nucleotide insertions and deletions, short tandem repeats. Short Genetic Variations may be common, thus representing true polymorphisms, or they may be rare. Some rare human entries have additional information associated with them, including disease associations (from ClinVar [2]), genotype information and allele origin, as some variations are somatic rather than germline events.



Short nucleotide variation data can be accessed via the SNP homepage and EUtils API:

www.ncbi.nlm.nih.gov/snp/ and www.ncbi.nlm.nih.gov/projects/SNP/SNPUtils.htm

VCF files and database bcp files are available for download through FTP and Aspera client at:

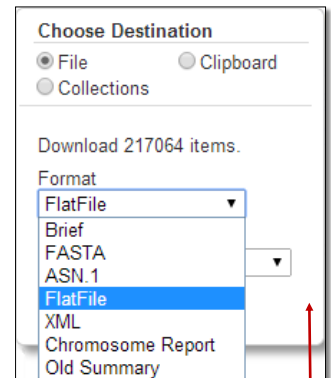
<ftp.ncbi.nlm.nih.gov/snp/> and www.ncbi.nlm.nih.gov/public/?snp/organisms/

SNP data can also be accessed interactively through Variation Viewer:

www.ncbi.nlm.nih.gov/variation/view/

For information on how to find known SNPs in a sequence, refer to this handout:

ftp.ncbi.nlm.nih.gov/pub/factsheets/HowTo_Finding_SNP_by_BLAST.pdf



Searching for and Displaying SNP Records

You can search for variations on the dbSNP homepage by typing a query term in the search box and clicking the Search button (A). You can also use the Advanced (B) page to create complex queries to produce more precise results. The search below, "hfe[Gene] AND human[orgn]", retrieves variations mapped to the human HFE gene. You can use options in the Display settings popup (C) to change the number of records displayed or sort retrieved variations in a different order. You can further narrow down retrieved variations by selecting filters present in the left column (D), or save them to a local file using the Send to (E) option. Use links to separate displays to see gene-centric listings (GeneView, F), graphical presentation under the context of genome or mRNA sequences (via HGVS names, G), or gene-centric display in a genomic context (Varview, H). Using the "Find related data" portlet (I), you can retrieve related entries from other NCBI databases for the set of variations in the display.

dbSNP Search Results for query: human[orgn] AND HFE[Gene]

Search: Search

Display Settings: Summary, 20 per page, Sorted by SNP_ID

Results: 1 to 20 of 841

1. rs1799945 [Homo sapiens]

2. rs1800562 [Homo sapiens]

3. rs1800562 [Homo sapiens]

4. rs1800562 [Homo sapiens]

5. rs1800562 [Homo sapiens]

6. rs1800562 [Homo sapiens]

7. rs1800562 [Homo sapiens]

8. rs1800562 [Homo sapiens]

9. rs1800562 [Homo sapiens]

10. rs1800562 [Homo sapiens]

11. rs1800562 [Homo sapiens]

12. rs1800562 [Homo sapiens]

13. rs1800562 [Homo sapiens]

14. rs1800562 [Homo sapiens]

15. rs1800562 [Homo sapiens]

16. rs1800562 [Homo sapiens]

17. rs1800562 [Homo sapiens]

18. rs1800562 [Homo sapiens]

19. rs1800562 [Homo sapiens]

20. rs1800562 [Homo sapiens]

Find related data

Database: Select

Find it

Search d

"Homo sapiens"

AND HFE[Gene]

Search

Recent a

Q human[orgn] AND HFE[Gene]

Format

Summary

Items per page

5

10

20

50

100

200

Sort by

Default order

Organism

SNP_ID

Success Rate

Heterozygosity

Chromosome Base Position

Apply

The Reference SNP Cluster Report

The Reference SNP Cluster Report linked from rsID ([rs1800730](#), shown in sections below and on p.3) provides details of a variation record. The report contains a summary of the allele (**A**), a link to the gene-centric display through the VarView icon (**B**, see p.4), mapping information in Human Genome Variation Society (HGVS) nomenclature (**C**), and minor allele frequencies (MAF, **D**) from various studies. The Integrated Maps table provides the genomic mapping details with the chromosomal coordinates (**E**) link to the same gene-centric display VarView icon provides. The magnifying glass (**F**) points to the 1000 Genomes Browser and provides genotyping details, if the variant this rsID represents is also called by that project.

For a summary of SNPs mapped to the gene, you can click the **Go** button (**G**) in the GeneView section to activate the **SNP: GeneView** display (p. 4).

The Gene Model(s) table below lists coordinates and changes on transcripts and proteins (**H**). The graphical panel (**I**) further below presents variants with various characteristics in different tracks (**J**) under the context of genome annotation. Individual variants are hyperlinked to provide additional details (**K**, activated upon hover).

Reference SNP (refSNP) Cluster Report: rs1800730			** With other allele **
RefSNP	Allele	HGVS Names	
Organism: human (<i>Homo sapiens</i>)	A	CM000668.2:g.26090957A>T	
Molecule Type: Genomic	C	NC_000006.11:g.26091185A>T	
Created/Updated in build: 89/151	Variation Class: SNV: single nucleotide variation	NC_000006.12:g.26090957A>T	
Map to Genome Build: 108/Weight 1	RefSNP Alleles: A/T (FWD)	NG_008720.2:g.8677A>T	
Validation Status:	Allele Origin: A:germline T:germline	NM_000410.3:c.193A>T	
Citation: PubMed LitVar NEW	Ancestral Allele: A	NM_001300749.1:c.193A>T	
	Variation Viewer: B	NM_139003.2:c.193A>T	
	Clinical Significance: D	NM_139004.2:c.193A>T	
		NM_139006.2:c.193A>T	
		NM_139007.2:c.77-357A>T	
		NM_139008.2:c.77-357A>T	
		NM_139009.2:c.124A>T	

Integrated Maps (Hint: click on 'Chr F' to see variant in the new NCBI variation viewer)										
Assembly	Annotation Release	Chr	Chr Pos	Contig	Contig Pos	SNP to Chr	Contig allele	Contig to Chr	Neighbor SNP	Map Method
GRCh38.p7	108	6	26090957	NT_007592.16	26030957	Fwd	A	Fwd	view	mapup
GRCh37.p13	105	6	26091185	NT_007592.15	26031185	Fwd	A	Fwd	view	blast

GeneView

GeneView via analysis of contig annotation: [HFE](https://go.usa.gov/xUHsc) *hemochromatosis*

[View more variation on this gene \(click to hide\).](#)

☒ Clinical Source: ☐ in gene region ☐ cSNP ☐ has frequency ☐ double hit

Primary Assembly Mapping

Assembly	SNP to Chr	Chr	Chr position	Contig	Contig position	Allele
GRCh38.p7	Fwd	6	26090957	NT_007592.16	26030957	A

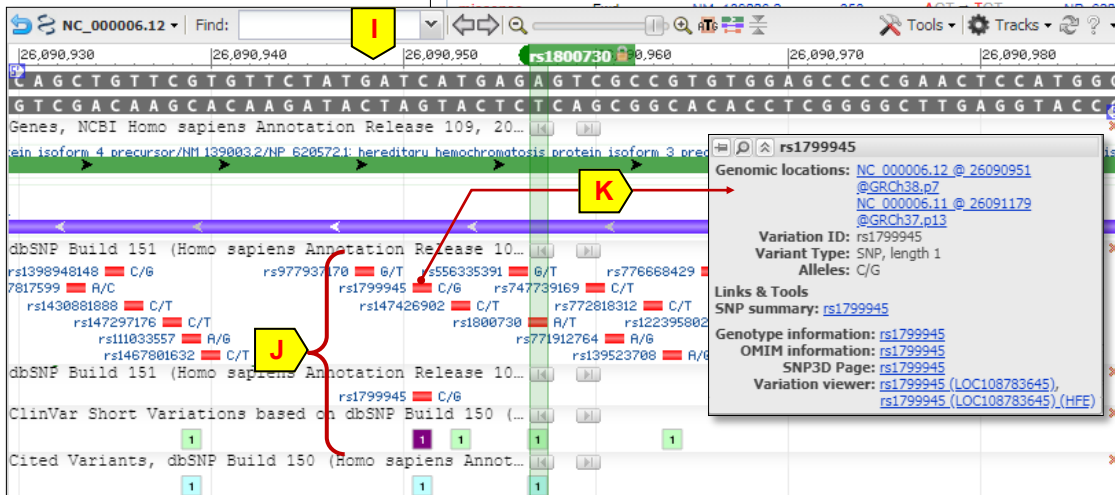
RefSeqGene Mapping

RefSeqGene	Gene (ID)	SNP to RefSeqGene	Position	Allele
NG_008720.2	HFE (3077)	Fwd	8677	A

Gene Model(s)

Function	mRNA			Protein			
	SNP to mRNA	Accession	Position	Allele change	Accession	Position	Residue change
missense	Fwd	NM_000410.3	353	AGT => TGT	NP_000401.1	65	S [Ser] => C [Cys]
missense	Fwd	NM_001300749.1	353	AGT => TGT	NP_001287678.1	65	S [Ser] => C [Cys]
missense	Fwd	NM_139003.2	353	AGT => TGT	NP_620572.1	65	S [Ser] => C [Cys]
missense	Fwd	NM_139004.2	353	AGT => TGT	NP_620573.1	65	S [Ser] => C [Cys]
	Fwd	NM_139006.2	353	AGT => TGT	NP_620575.1	65	S [Ser] => C [Cys]
	Fwd	NM_139007.2	353	AGT => TGT	NP_620576.1	65	S [Ser] => C [Cys]
	Fwd	NM_139008.2	353	AGT => TGT	NP_620577.1	65	S [Ser] => C [Cys]
	Fwd	NM_139009.2	353	AGT => TGT	NP_620578.1	65	S [Ser] => C [Cys]

<



The Submitter records table (**L**) lists alleles and flanking sequences from submitter SNPs (ssIDs) included in this reference SNP cluster. The ssIDs (**M**) link to submitter records with additional details.

Submitter records for this RefSNP Cluster										
The submission ss244317424 has the longest flanking sequence of all cluster members and was used to instantiate sequence for rs1800730 during BLAST analysis for the current build.										
NCBI Assay ID	Handle/Submitter ID	Validation Status	ss to rs Orientation /Strand	Alleles	5' Near Seq 30 bp	3' Near Seq 30 bp	Entry Date	Update Date	Build Added	Molecule Type
ss2420855	HGBASE SNP000007282		fwd/T	A/T	gctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactcc	11/07/00	10/10/03	89	Genomic
ss68969473	PERLEGEN PGP04777602		fwd/I	A/T	gaccagctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactccatggg	01/30/07	08/14/07	127	Genomic
ss160462894	ILLUMINA HumanOmni1-Quad_v1-0_B		fwd/I	A/T	gaccagctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactccatggg	08/04/09	10/02/09	131	Genomic
ss233370756	1000GENOMES pilot_1_CEU_2975385_chr6_261		fwd/I	A/T	gaccagctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactccatggg	05/01/10	05/01/10	132	Genomic
ss244317424	2010_April_001_075_HFE_235200_0003...		fwd/I	A/T	gaccagctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactccatggg	06/16/10	06/16/10	132	Genomic
ss28828992	OMIM CURATED-RECORDS 6335		fwd/I	A/T	gaccagctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactccatggg	12/21/10	12/21/10	133	Genomic
ss342203118	NHLBI ESP ESP2500-chr6-26091185		fwd/I	A/T	gaccagctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactccatggg	03/25/11	03/26/11	134	Genomic
ss410868034	ILLUMINA Cardio-Metabo_Chip_A_chr6_26199164...		fwd/I	A/T	gaccagctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactccatggg	06/07/11	06/07/11	135	Genomic
ss481067367	ILLUMINA HumanOmni1-Quad_v1-0		fwd/I	A/T	gaccagctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactccatggg	01/30/12	01/31/12	137	Genomic
ss490921024	1000GENOMES 20110521_exome_428822_chr6_260...		fwd/I	A/T	gaccagctgttcgtgttctatgatcatgag	gtcgcctgtgtggagcccccgaactccatggg	02/10/12	02/21/12	137	Genomic

The Reference SNP Cluster Report (cont.)

The **FASTA Sequence** section shows the sequences flanking the variation. It uses the exemplar submitter record to represent the variation by presenting the sequence as the 5'-(**A**) and 3'-(**B**) flanking sequences, with the allele (**C**) in the middle. The **Population Diversity** section (**D**) sums up available genotype and allele frequency information for various populations from different studies. More genotype details are available through the 1000 Genomes browser link (**E**).

Fasta sequence (Legend)

```
>gnl|dbSNP|rs1800730|allelePos=251|totalLen=501|taxid=9606|snpclass=1|alleles='A/T'|mol=Ge
CAGGACTGCA ACTCACCCTT CACAAAATGA GGACCAGACA CAGCTGATGG TATGAGTTGA TGCAGGTGTG
TGGAGGCTCA ACATCCTGCT CCCTCTCTAC TACACATGGT TAAGGCCTGT TGCTCTGTCT CCAGGTTTCA
ACTCTCTGCA CTACCTCTTC ATGGGTGCTT CAGAGCAGGA CTTTGGTCTT TCCTTGTITG AAGCTTTGGG
CTACGTCTCA GACCAGCTGT TCGTGTCTA TGATCATGAG
W
GTGCGCTCTT GGAGCCCGGA ATCCCATGGG TTCCAGTAG AATTTCAAGC CAGATGTGGC TGCAGCTGAG
TCAGAGTCTG AAAGGGTGGG ATCATCATGT CACTGTTGAC TTCTGGACTA TTATGGAAAA TCACAACCAC
AGCAAGGGTA TGTGGAGAGG GGGCCTCACC TTCTGAGGT TGTGAGACT TTTTCATCTT TCATGCACTT
TGAAGGAAC AGCTGGAAGT CTGAGGTCTT GTGGGAGCAG
```

Population Diversity (Alleles in RefSNP orientation) . See additional population frequency from 1000Genomes [here]

Sample Ascertainment				Genotype Detail				Alleles	
ss#	Population	Individual Group	Chrom. Sample Cnt.	Source	A/A	A/T	T/T	HWP	A T
ss1319404492	EAS		1008	AF					1.00000000
	EUR		1006	AF					0.98409998 0.01590000
	AFR		1322	AF					1.00000000
	AMR		694	AF					0.99570000 0.00430000
	SAS		978	AF					0.99899995 0.00100000
ss1688187409	ExAc_Aggregated_Populations		121412	AF					0.98991036 0.01008961
ss233370756	pilot_1_CEU_low_coverage_panel		120	AF					0.96666664 0.03333334
ss342203118	ESP_Cohort_Populations		4552	GF	0.97803164	0.02196836		0.65472100	0.98901582 0.01098418
ss491881981	CSAqilent		1323	GF	0.98299998	0.01700000		1.00000000	0.99150002 0.00850000

Other Ways to Access Data from dbSNP

The SNP database is fully integrated with the Entrez system, enabling the access of variation data through links present in records from other NCBI databases.

For example, you can use the **SNP: GeneView** or **Variation Viewer** link (**F**) found in the **Related information** section of a Gene database record to see a summary list of variations mapped to that gene. You can project variations mapped to a segment of the RefSeq genomic or a mRNA record (with NT_, NG_, NW_ or NM_ accessions) by using the **Customize view** (**G**) menu in the upper right hand corner of the sequence record, simply check the SNPs checkbox and click **Update View** (**H**) to activate the selection.

Customize view

Basic Features

- ☒ Default features
- ☐ Gene, RNA, and CDS features only

Features added by NCBI

- ☒ 65 SNPs

Display options

- ☒ Show sequence
- ☐ Show reverse complement

Update View

```
exon      1..236
          /gene="HFE"
          /gene_synonym="HFE1; HH; HLA-H; MVCD7; TFQTL2"
          /inference="alignment:Splign"
          /number=1
          34
          /gene="HFE"
          /gene_synonym="HFE1; HH; HLA-H; MVCD7; TFQTL2"
          /replace="c"
          /replace="g"
          /db_xref="dbSNP:62625316"
          49
          /gene="HFE"
          /gene_synonym="HFE1; HH; HLA-H; MVCD7; TFQTL2"
          /replace="c"
          /replace="t"
          /db_xref="dbSNP:62625317"
```

Related information

Order cDNA clone

3D structures

ClinVar

Conserved Domains

dbVar

Full text in PMC

Full text in PMC_nucleotide

Functional Class

GAP

Gene neighbors

Genome

GEO Profiles

GTR

HomoloGene

MedGen

Nucleotide

OMIM

Probe

Protein

PubChem Compound

PubChem Substance

PubMed

PubMed (GeneRIF)

PubMed (OMIM)

PubMed(nucleotide/PMC)

RefSeq Proteins

RefSeq RNAs

RefSeqGene

Related gene-specific medical variations

SNP

SNP: GeneView

Variation Viewer

dbSNP also integrates disease-related nucleotide variations that were reported in literature and cited in rsID format, collected by OMIM, or submitted to ClinVar. The table below is the **Allelic Variant** display for OMIM record 613609, which cites the rsIDs in the dbSNP column (**I**).

613609

HFE GENE; HFE

Allelic Variants (11 Selected Examples) :

All ClinVar Variants

Number	Phenotype	Mutation	dbSNP	ExAC	ClinVar
.0001	HEMOCHROMATOSIS, TYPE 1 PORPHYRIA CUTANEA TARDA, SUSCEPTIBILITY TO, INCLUDED PORPHYRIA VARIEGATA, SUSCEPTIBILITY TO, INCLUDED HEMOCHROMATOSIS, JUVENILE, DIGENIC, INCLUDED ALZHEIMER DISEASE, SUSCEPTIBILITY TO, INCLUDED TRANSFERRIN SERUM LEVEL QUANTITATIVE TRAIT LOCUS 2, INCLUDED MICROVASCULAR COMPLICATIONS OF DIABETES, SUSCEPTIBILITY TO, 7, INCLUDED	HFE, CYS282TYR	[rs1800562]	-	[RCV0002108]
.0002	HEMOCHROMATOSIS, TYPE 1 MICROVASCULAR COMPLICATIONS OF DIABETES, SUSCEPTIBILITY TO, 7, INCLUDED	HFE, HIS63ASP	[rs1799945]	[rs1799945]	[RCV0000000]
.0003	HEMOCHROMATOSIS, TYPE 1	HFE, SER65CYS	[rs1800730]	-	[RCV0002907]
.0004	HFE INTRONIC POLYMORPHISM	HFE, 5569G-A	[rs1800758]	[rs1800758]	[RCV0000000]
.0005	HFE POLYMORPHISM	HFE, VAL53MET	[rs28934889]	-	[RCV0000000]
.0006	HFE POLYMORPHISM	HFE, VAL59MET	[rs111033557]	-	[RCV0000000]
.0007	HEMOCHROMATOSIS, TYPE 1	HFE, GLN127HIS	[rs28934595]	-	[RCV0000000]
.0008	HEMOCHROMATOSIS, TYPE 1	HFE, ARG330MET	[rs111033558]	-	[RCV0000000]

The SNP:GeneView Display

The SNP:GeneView display tabulates variations mapped to splice variants of a particular gene. At the top, it lists all annotated splice variants (A) of the gene. The splice variant, whose variations are shown, is highlighted in yellow (B). The default setting shows only the non-clinical coding variations. Check "Clinical Source" and "in gene region" options, then click "Reresh" (C) to see the complete list. The table arranges mapped variants by their chromosomal coordinates (D) and color-codes them by their function: **white** for "in gene region" (E), **orange** for UTR (F), **green** for synonymous (G), **red** for non-synonymous (H), **blue** for frame-shift (I), **purple** for splicing site (J), and **yellow** for intron (K). The MAF (L) column lists the global minor allele frequencies from the 1000 Genomes project.

Variation Viewer

The GeneView display contains a link (M) to an interactive display in the Variation Viewer (N), which can display variation mapping and molecular consequences within the GRCh37 or GRCh38 context for more focused examination - by correlating a variation and its molecular consequences in the data table with its genomic context in the graphical display (O). Filters in the left hand column (not shown) are available to selectively display variants of interest.

More information on this tool is available online as a video tutorial, in a fact-sheet, as well as in the more detailed online help:

Variation Viewer factsheet
Online video tutorial
Variation Viewer help

SNP linked to Gene (geneID:3077) Via Contig Annotation

The SNP GeneView page only reports human variation on GRCh38. A new [Variation Viewer](#) is available to view the gene HFE variations in [GRCh37p13](#) or [GRCh38](#), and will replace SNP GeneView later this year. Please visit the [Help Page](#) or [YouTube](#) for available features and send your comments and suggestions to [NCBI helpdesk](#).

Send [on all gene models to Batch Query](#) Download all rs# to file

Gene model (mRNA alignment) information from genome sequence

Total gene model (contig mRNA transcript): 11

mRNA	transcript	protein	mRNA orientation	Contig	Contig Label	List SNP
NM_000410.3	plus strand	NP_000401.1	forward	NT_007592.16	GRCh38.p7	<- currently shown
XM_011514543.2	plus strand	XP_011512845.1	forward	NT_007592.16	GRCh38.p7	View snp on GeneModel
NM_139011.2	plus strand	NP_620580.1	forward	NT_007592.16	GRCh38.p7	View snp on GeneModel
NM_139010.2	plus strand	NP_620579.1	forward	NT_007592.16	GRCh38.p7	View snp on GeneModel

Model transcript: GRCh38.p7 NT_007592.16 NM_000410.3 NP_000401.1 forward plus strand 755, all

Region	Chr.	mRNA pos	dbSNP rs#	Heterozygosity	Validation	MAF	Allele origin	3D	Clinically Associated	Clinical Significance	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos	PubMed
5' near gene	26085282		rs72834669	0.040		0.0202					5' near gene					
	26085291		rs146329216	0.001		0.0004					5' near gene					
	26087393	113	rs41266793	0.002		0.0012					5' UTR	A				
											5' UTR	G				
	26091501	688	rs779438836	0.000							synonymous	G	Ala [A]	3	176	
											contig reference	C	Ala [A]	3	176	
	26091518	705	rs549729183	0.000		0.0002					missense	G	Cys [C]	2	182	
											contig reference	A	Tyr [Y]	2	182	
	26091519	706	rs765804978	0.000							frame shift	-	Gly [G]	3	183	
											contig reference	CC	Leu [L]	3	183	
	26091590		rs773443949	0.000							donor					
	26091602		rs377202967	0.000		0.0002					intron					

Variation Viewer Homo sapiens: GRCh38.p7 (GCF_000001405.33) Chr 6 (NC_000006.12): 26,090,938 - 26,090,987

Reset All Share this page FAQ Help You Tube

Region: LOC108783645 NR_144383.1

Gene: Transcript

Exons: click an exon above to zoom in, mouse over to see details

NC_000006.12: 26M..26M (69bp)

rs1800730

ClinVar Short Variations based on dbSNP Build 150 (Homo sapiens Annotation Release 108)

dbSNP Build 151 (Homo sapiens Annotation Release 108)

rs1398948148 C/G rs777817599 A/C rs1430861888 C/T rs147297176 C/T rs11833557 A/G rs1467801632 C/T rs977997170 G/T rs1800730 A/T rs7799945 C/G rs171912764 A/G rs147426902 C/T rs556335391 G/T rs776668429 A/G rs747739169 C/T rs139523788 A/G/T rs1314720488 A/G rs1263353185 A/C rs752596302 C/T rs62625342 C/T rs945769842 C/G rs7643438 rs1177

rs556335391 26,090,956 single nucleotide variant LOC108783645 and 1 more non coding transcript variant, missense variant, intron variant T = 0.0004 T = 1.6e-5

rs1800730 26,090,957 single nucleotide variant LOC108783645 and 1 more intron variant, missense variant, non coding transcript variant Pathogenic T = 0.0040 T = 0.0111 T = 0.0101 16

Alleles associated with rs1800730

Variant allele	Transcript change	RefSeq	Protein change	Molecular consequence	Condition	Most severe clinical significance	Submitters	Highest review status	Last evaluated
T	c.193A>T	NM_000410.3	Ser65Cys	missense variant	Hemochromatosis type 1 and 1 more	Pathogenic	5	criteria provided, conflicting interpretations	Jun 21, 2016
T	c.193A>T	NM_001300749.1	Ser65Cys	missense variant	Hemochromatosis type 1 and 1 more	Pathogenic	5	criteria provided, conflicting interpretations	Jun 21, 2016
T	c.193A>T	NM_139003.2	Ser65Cys	missense variant	Hemochromatosis type 1 and 1 more	Pathogenic	5	criteria provided, conflicting interpretations	Jun 21, 2016

https://ftp.ncbi.nih.gov/pub/factsheets/Factsheet_Variation_Viewier.pdf

<https://www.youtube.com/watch?v=rnWZ9MFBwUM>

<https://www.ncbi.nlm.nih.gov/variation/view/help/>